PDHX gene

pyruvate dehydrogenase complex component X

Normal Function

The *PDHX* gene provides instructions for making a protein called E3 binding protein, which is part of a large group of proteins known as the pyruvate dehydrogenase complex. This complex is made up of several enzymes, including one called E3, and other proteins. E3 binding protein attaches E3 to the complex and provides the correct structure for the complex to perform its function.

The pyruvate dehydrogenase complex plays an important role in the pathways that convert the energy from food into a form that cells can use. This enzyme converts a molecule called pyruvate, which is formed from the breakdown of carbohydrates, into another molecule called acetyl-CoA. This conversion is essential to begin the series of chemical reactions that produces adenosine triphosphate (ATP), the cell's main energy source.

Health Conditions Related to Genetic Changes

Leigh syndrome

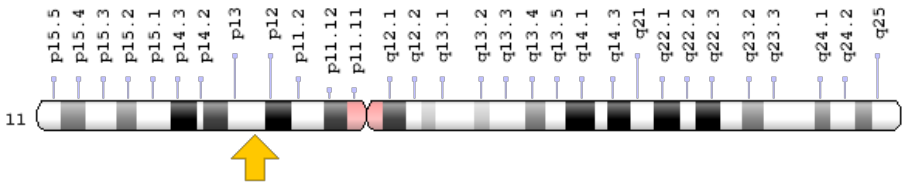
pyruvate dehydrogenase deficiency

Mutations in the *PDHX* gene cause pyruvate dehydrogenase deficiency in a small number of people. This condition is characterized by a potentially life-threatening buildup of a chemical called lactic acid in the body (lactic acidosis), delayed development, and neurological problems. *PDHX* gene mutations associated with pyruvate dehydrogenase deficiency result in the complete absence of E3 binding protein. Loss of this protein impairs the binding of the E3 enzyme to the pyruvate dehydrogenase complex, which leads to a reduction of the complex's activity. With decreased function of this complex, pyruvate builds up and is converted, in another chemical reaction, to lactic acid, causing lactic acidosis. In addition, the production of cellular energy is diminished. The brain, which is especially dependent on this form of energy, is severely affected, resulting in the neurological problems associated with pyruvate dehydrogenase deficiency.

Chromosomal Location

Cytogenetic Location: 11p13, which is the short (p) arm of chromosome 11 at position 13

Molecular Location: base pairs 34,915,829 to 34,996,128 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- dihydrolipoamide dehydrogenase-binding protein of pyruvate dehydrogenase complex
- DLDBP
- E3BP
- lipoyl-containing pyruvate dehydrogenase complex component X
- ODPX_HUMAN
- OPDX
- proX
- pyruvate dehydrogenase complex, component X
- pyruvate dehydrogenase complex, E3-binding protein subunit
- pyruvate dehydrogenase complex, lipoyl-containing component X
- pyruvate dehydrogenase protein X component, mitochondrial

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): The Formation of Acetyl Coenzyme A from Pyruvate
  https://www.ncbi.nlm.nih.gov/books/NBK22427/#A2376
- Biochemistry (fifth edition, 2002): The Pyruvate Dehydrogenase Complex Is Regulated Allosterically and by Reversible Phosphorylation
  https://www.ncbi.nlm.nih.gov/books/NBK22347/#A2410
- Molecular Biology of the Cell (fourth edition, 2002): Sugars and Fats Are Both Degraded to Acetyl CoA in Mitochondria
  https://www.ncbi.nlm.nih.gov/books/NBK26882/#A300
- Molecular Cell Biology (fourth edition, 2000): Mitochondrial Oxidation of Pyruvate Begins with the Formation of Acetyl CoA
  https://www.ncbi.nlm.nih.gov/books/NBK21624/#A4352

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28PDHX%5BTIAB%5D%29+OR+%28%28E3BP%5BTIAB%5D%29+OR+%28DLDBP%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- PYRUVATE DEHYDROGENASE COMPLEX, COMPONENT X
  http://omim.org/entry/608769

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PDHX.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PDHX%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=21350
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/8050
- UniProt
  http://www.uniprot.org/uniprot/O00330

Sources for This Summary

- Biochemistry (fifth edition, 2002): The Formation of Acetyl Coenzyme A from Pyruvate
  https://www.ncbi.nlm.nih.gov/books/NBK22427/#A2376
- Brown RM, Head RA, Brown GK. Pyruvate dehydrogenase E3 binding protein deficiency. Hum Genet. 2002 Feb;110(2):187-91. Epub 2002 Jan 22.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11935326
- Brown RM, Head RA, Morris AA, Raiman JA, Walter JH, Whitehouse WP, Brown GK. Pyruvate dehydrogenase E3 binding protein (protein X) deficiency. Dev Med Child Neurol. 2006 Sep;48(9): 756-60.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16904023
- OMIM: PYRUVATE DEHYDROGENASE COMPLEX, COMPONENT X
  http://omim.org/entry/608769
- Patel MS, Korotchkina LG, Sidhu S. Interaction of E1 and E3 components with the core proteins of the human pyruvate dehydrogenase complex. J Mol Catal B Enzym. 2009 Nov 1;61(1-2):2-6.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20160912
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2770179/
- Smolle M, Prior AE, Brown AE, Cooper A, Byron O, Lindsay JG. A new level of architectural complexity in the human pyruvate dehydrogenase complex. J Biol Chem. 2006 Jul 14;281(28): 19772-80. Epub 2006 May 5.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16679318
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3954457/

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